CLAIMS

1. A method for detecting the presence or absence of a mutation associated with hypertrophic cardiomyopathy, comprising:

amplifying ß cardiac myosin heavy-chain DNA forming an amplified product; and

detecting the presence or absence of a mutation associated with hypertrophic cardiomyopathy in the amplified product.

2. The method of claim 1 wherein the hypertrophic cardiomyopathy is familial hypertrophic cardiomyopathy.

3. The method of claim 1 wherein the hypertrophic cardiomyopathy is sporadic hypertrophic cardiomyopathy.

4. The method of claim 2 wherein the mutation associated with hypertrophic cardiomyopathy is a point mutation.

5. The method of claim 4 wherein the point mutation is a missense mutation.

6. The method of claim 1 wherein the mutation associated with hypertrophic cardiomyopathy is a small alteration in the amplified DNA.

7. The method of claim 1 wherein the ß cardiac myosin heavy-chain DNA is cDNA reversed transcribed from RNA.

8. The method of claim 7 wherein the RNA is obtained from nucleated blood cells.

9. The method of claim 1 wherein the presence or absence of the mutation associated with hypertrophic cardiomyopathy is detected by combining the amplified product with an RNA probe completely hybridizable to normal ß cardiac myosin heavy-chain DNA forming a hybrid double strand having an RNA and DNA strand, the hybrid double strand having an unhybridized portion of the RNA strand at any portion corresponding to a hypertrophic cardiomyopathy associated mutation in the DNA strand; and

detecting the presence or absence of an unhybridized portion of the RNA strand as an indication of the presence or absence of a hypertrophic cardiomyopathy associated mutation in the corresponding portion of the DNA strand.

10. The method of claim 2 wherein the presence or absence of the mutation associated with familial hypertrophic cardiomyopathy is detected by combining the amplified product with an RNA probe completely hybridizable to normal ß cardiac myosin heavy-chain DNA forming a hybrid double strand having an RNA and DNA strand, the hybrid dobule strand having an unhybridized ribonucleotide of the RNA strand at any portion corresponding to a familial hypertrophic cardiomyopathy associated point mutation in the DNA strand;

contacting the hybrid double strand with an agent capable of digesting an unhybridized portion of the RNA strand; and

detecting the presence or absence of an unhybridized ribonucleotide of the RNA strand as an indication of the presence or absence of a familial hypertrophic cardiomyopathy associated point mutation in the corresponding deoxyribonucleotide of the DNA strand.

11. The method of claim 1 wherein the β cardiac myosin heavy-chain DNA is amplified using a polymerase chain reaction.

12. The method of claim 11 wherein the polymerase chain reaction is a nested polymerase chain reaction.

13. A method for diagnosing familial hypertrophic cardiomyopathy comprising:

obtaining a sample of β cardiac myosin heavy-chain DNA derived from a subject being tested for hypertrophic cardiomyopathy; and

diagnosing the subject for familial hypertrophic cardiomyopathy by detecting the presence or absence of a familial hypertrophic cardiomyopathy-causing point mutation in the β cardiac myosin heavy-chain DNA as an indication of the disease.

14. The method of claim 13 wherein the β cardiac myosin heavy-chain DNA is cDNA reverse transcribed from RNA obtained from the subject's nucleated blood cells.

15. The method of claim 13 further comprising amplifying the ß cardiac myosin heavy-chain DNA prior to the diagnosis step.

16. The method of claim 15 wherein an exon suspected of containing the familial hypertrophic cardiomyopathy-causing point mutation is selectively amplified.

17. The method of claim 13 wherein the point mutation is selected from the group consisting of Arg249Gln, Arg403Gln, Arg453Cys, Gly584Arg, Val606Met, Glu924Lys, and Glu949Lys.

18. A non-invasive method for diagnosing hypertrophic cardiomyopathy, comprising:

obtaining a blood sample from a subject being tested for hypertrophic cardiomyopathy;

isolating ß cardiac myosin heavy-chain RNA from the blood sample; and

diagnosing the subject for hypertrophic cardiomyopathy by detecting the presence or absence of a hypertrophic cardiomyopathy-associating mutation in the RNA as an indication of the disease.

19. The method of claim 18 wherein the presence or absence of a hypertrophic cardiomyopathy-associated mutation in the RNA is detected by preparing ß cardiac myosin heavy-chain cDNA from the RNA forming ß cardiac myosin heavy-chain DNA and detecting mutations in the DNA as being indicative of mutations in the RNA.

20. The method of claim 18 further comprising amplifying the ß cardiac myosin heavy-chain DNA prior to detecting a hypertrophic cardiomyopathy-associated mutations in the DNA.

21. The method of claim 18 wherein the hypertrophic cardiomyopathy is familial hypertrophic cardiomyopathy.

22. The method of claim 18 wherein the hypertrophic cardiomyopathy is sporadic hypertrophic cardiomyopathy.

23. The method of claim 18 further comprising evaluating the subject for clinical symptoms associated with familial hypertrophic cardiomyopathy.

24. A method for detecting the presence or absence of a disease associated mutation in a DNA sequence, comprising:

amplifying a DNA sequence suspected of containing a disease-associated mutation forming an amplified product;

combining the amplified product with an RNA probe completely hybridizable to a normal DNA sequence associated with the disease forming a hybrid double strand having an RNA and DNA strand, the hybrid double strand having an unhybridized portion of the RNA strand at a portion corresponding to a disease-associated mutation in the DNA strand; and

detecting the presence or absence of an unhybridized portion of the RNA strand as an indication of the presence or absence of a disease-associated mutation in the corresponding portion of the DNA strand.

25. The method of claim 24 wherein the disease-associated mutation is a point mutation in the DNA strand.

26. The method of claim 24 wherein the disease-associated mutation is a small alteration in the DNA strand.

27. The method of claim 24 wherein the presence or absence of an unhybridized portion of the RNA strand is detected by contacting the hybrid double strand with an agent capable of digesting an unhybridized portion of the RNA strand, denaturing the hybrid double strand, separating the RNA fragments by size, and comparing the fragments of RNA resulting from portions of the RNA strand being digested by the agent to RNA fragments representative of normal RNA.

28. The method of claim 24 further comprising sequencing a portion of DNA corresponding to an unhybridized portion of the RNA strand to identify the sequence of a disease associated mutation.

29. The method of claim 24 wherein the presence or absence of more than one unhybridized portion of the RNA strand are detected as an indication of the presence or absence of more than one disease associated mutation in the corresponding portions of the DNA strand.

30. The method of claim 24 whereins the DNA sequence suspected of containing a disease-associated mutation is amplified using a polymerase chain reaction.

31. The method of claim 30 wherein the polymerase chain reaction is a nested polymerase chain reaction.

32. A method for determining the estimated life expectancy of a person having familial hypertrophic cardiomyopathy, comprising:

obtaining ß cardiac myosin DNA derived from a subject having familial hypertrophic cardiomyopathy;

detecting a familial hypertrophic cardiomyopathy-causing point mutation in the ß cardiac myosin DNA;

classifying the type of familial hypertrophic cardiomyopathy-causing point mutation; and

estimating the life expectancy of the subject using a Kaplan-Meier curve for the classified type of familial hypertrophic cardiomyopathy-causing point mutation.

33. A kit useful for diagnosing hypertrophic cardiomyopathy, comprising:

a first container holding an RNA probe completely hybridizable to the ß cardiac myosin heavy chain DNA; and

a second container holding primers useful for amplifying ß cardiac myosin heavy-chain DNA.

34. A kit of claim 33 further comprising a third container holding an agent for digesting unhybridized RNA.

35. The kit of claim 33 further comprising instructions for using the components of the kit to detect the presence or absence of hypertrophic cardiomyopathy-associated point mutations in amplified ß-cardiac myosin heavy-chain DNA.

36. An RNA probe comprising ribonucleotides arranged in a sequence which is complementary to at least a portion of ß-cardiac myosin heavy-chain DNA.

37. A set of DNA oligonucleotide primers for amplifying ß-cardiac myosin heavy-chain DNA comprising, at least two oligonucleotides capable of amplifying ß-cardiac myosin heavy-chain DNA.

38. The set of primers of claim 37 having four oligonucleotides.